PHOX2A gene

paired like homeobox 2a

Normal Function

The *PHOX2A* gene provides instructions for making a protein that is found in the nervous system. This protein acts early in development to help promote the formation of nerve cells (neurons) and regulate the process by which the neurons mature to carry out specific functions (differentiation).

Most of researchers' knowledge about the PHOX2A protein comes from studies in other animals. From these studies, it is clear that the protein plays a critical role in the development of the autonomic nervous system, which controls involuntary body functions such as breathing, blood pressure, heart rate, and digestion. The PHOX2A protein is also involved in the formation of certain nerves in the head and face. Specifically, it appears to be critical for the development and function of cranial nerves III and IV, which emerge from the brain and control many of the muscles that surround the eyes (extraocular muscles). These muscles direct eye movement and determine the position of the eyes.

Health Conditions Related to Genetic Changes

congenital fibrosis of the extraocular muscles

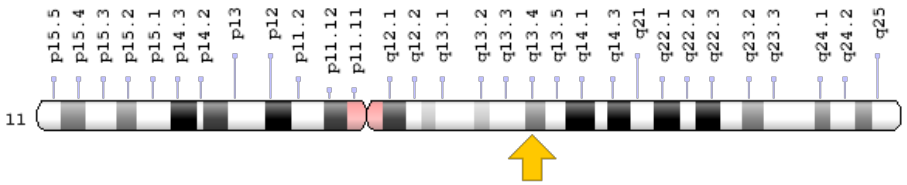
At least four mutations in the *PHOX2A* gene can cause congenital fibrosis of the extraocular muscles. These mutations are responsible for a form of the disorder called CFEOM2, which has been identified in several families of Middle Eastern descent.

Most of the mutations that cause congenital fibrosis of the extraocular muscles result in the production of an abnormally short, nonfunctional version of the PHOX2A protein. In humans, a lack of this protein prevents the normal development of several cranial nerves and the extraocular muscles they control. Abnormal development and function of these muscles leads to the characteristic features of the disorder, including restricted eye movement and related problems with vision. Although the PHOX2A protein plays an important role in autonomic nervous system development, *PHOX2A* mutations do not seem to affect the function of this part of the nervous system.

Chromosomal Location

Cytogenetic Location: 11q13.4, which is the long (q) arm of chromosome 11 at position 13.4

Molecular Location: base pairs 72,239,077 to 72,244,176 on chromosome 11 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- aristaless homeobox homolog
- aristaless homeobox protein homolog
- ARIX
- arix homeodomain protein
- ARIX1 homeodomain protein
- CFEOM2
- FEOM2
- MGC52227
- NCAM2
- paired-like homeobox 2a
- paired mesoderm homeobox protein 2A
- PHX2A_HUMAN
- PMX2A

Additional Information & Resources

Educational Resources

- Neuroscience (2nd edition, 2001): The Actions and Innervation of Extraocular Muscles
  https://www.ncbi.nlm.nih.gov/books/NBK10793/

GeneReviews

- Congenital Fibrosis of the Extraocular Muscles
  https://www.ncbi.nlm.nih.gov/books/NBK1348

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28PHOX2A%5BTIAB%5D%29+OR+%28ARIX%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- ARISTALESS HOMEOBOX, DROSOPHILA, HOMOLOG OF
  http://omim.org/entry/602753

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_PHOX2A.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=PHOX2A%5Bgene%5D
- HGNC Gene Family: PRD class homeoboxes and pseudogenes
  http://www.genenames.org/cgi-bin/genefamilies/set/521
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=691
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/401
- UniProt
  http://www.uniprot.org/uniprot/O14813

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